



PARENT / GUARDIAN INFORMATION & CONSENT SHEET

This information is for dual parent and doctor participation.

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| HREC PROJECT NUMBER | 17/05/17/4.06 |
| RESEARCH PROJECT TITLE | The FaceMatch: Searching for a diagnosis project |
| PRINCIPAL RESEARCHER | Dr Tracy Dudding-Byth, Consultant Clinical Geneticist, Hunter New England Health Clinical Research Fellow |

The FaceMatch Project is generously funded by the
Newcastle Permanent Charitable Foundation.

The FaceMatch project is approved by
Hunter New England Health Ethics Committee
and supported by Hunter Medical Research Institute and
Hunter New England Health Research, Innovation & Partnerships.



We would like to invite your son/daughter to participate in **The FaceMatch: searching for a diagnosis project**.

This document provides an information statement about the project and access to an online consent form.

WHAT IS AN INFORMATION STATEMENT?

This information statement explains clearly and openly all the steps involved in participating in the FaceMatch Project. The information is to help you decide whether or not you would like your son/daughter to be enrolled in the project.

Before you decide if you want your son/daughter to take part or not, you may want to talk about the project with your family, a friend or a health care provider. You can also contact the FaceMatch team by email: info@FaceMatch.org.au

CONSENT PROCESS

By completing the consent process below, you are telling us that you:

- Understand what you have read.
- Had a chance to ask questions and received satisfactory answers.
- Consent to your child taking part in the project.

You can print a copy of this information and consent form to keep for your records.

WHAT IS THE BACKGROUND TO THIS RESEARCH PROJECT?

Understanding the genetic cause for an individual's ID can provide information for the management of the condition and reduce the sense of isolation for the family. It can also help parents make decisions about family planning. Understanding the genetic causes of ID is also the first step towards developing future treatments.

Moderate to severe intellectual disability (IQ less than 50) affects one in 200 children and 30-50 percent of these children have facial features which look different from the rest of their family. In developed countries, the most common cause of intellectual disability (ID) is genetic. In some cases, a doctor is able to make a genetic diagnosis based on a collection of recognisable facial features.

Improved genetic testing has made it possible to test an individual's entire genetic makeup (the genome) including all the known (approximately 1500) developmental genes in a single test.

However, a diagnosis is still only achieved approximately 30-40 percent of the time.

WHAT IS THE AIM OF THIS RESEARCH PROJECT?

The aim of the FaceMatch Project is to use IMAGUS computer face-matching technology to try and accurately match the faces of individuals with undiagnosed syndromic forms of intellectual disability.

By matching faces with similar features, we hope to match individuals with the same rare genetic conditions. We hope this will aid diagnosis and also lead to the discovery of new genes associated with ID.

The FaceMatch Project will also compare the faces of undiagnosed individuals with ID to those who have a confirmed genetic diagnosis.

The study hopes to find new genes involved in causing ID and improve the current rate of obtaining a diagnosis for families. The FaceMatch Project will offer international recruitment through a secure website.

WHO CAN PARTICIPATE IN THE RESEARCH PROJECT?

We will be inviting the following groups of people to participate.

1. Children or adults with moderate to severe intellectual disability who remain undiagnosed following a review by a clinical geneticist or medical specialist.
2. Children or adults who have a known genetic cause for their intellectual disability.

WHO IS FUNDING THIS RESEARCH?

The project is supported by a 2017 Hunter New England Health Clinical Research Fellowship and a 2018 Newcastle Permanent Charitable Foundation grant.

WHY IS MY SON/DAUGHTER BEING ASKED TO BE IN THIS RESEARCH PROJECT?

We are asking your son/daughter to take part in this project because;

- he/she has moderate to severe intellectual disability, but remains undiagnosed following a review by a clinical geneticist or medical specialist.
- he/she has been given a specific genetic diagnosis to account for his/her intellectual disability.

WHAT DOES PARTICIPATION IN THE RESEARCH INVOLVE?

You will be asked to provide information for your son/daughter's profile page. This will include uploading a photograph(s) of your son/daughter at different ages into the secure FaceMatch database.

You will be asked to provide the name of your son/daughter's clinical geneticist or medical specialist and a contact email for this nominated doctor. Tips on how to obtain this information will be provided.

The doctor you nominate will be asked to verify that your son/daughter is their patient and will be invited to provide relevant medical information about your child.

Every new image uploaded into the FaceMatch database will automatically be matched against all other photographs within the database.

Photographs identified as very similar (high-probability facial matches) will be reviewed by the FaceMatch research team and the doctors of the matching individuals, within the FaceMatch project.

Your nominated doctor will contact you if a high-probability facial match has been made.

WHAT ARE MY SON/DAUGHTER'S ALTERNATIVES TO TAKING PART?

Participation in this research project is voluntary. It is your choice to let your son/daughter take part in the project. You do not need to agree to participate if you do not want to.

If you give your consent and change your mind, you can withdraw from the project at any time by simply emailing our team at info@FaceMatch.org.au. You do not need to tell us the reason why you want to withdraw.

If you decide to leave the project, we will retain securely any de-identified photographs and information already collected unless you tell us not to. Whatever your decision, it will not affect any treatment or care your son/daughter receives from health services.

WHAT ARE THE POSSIBLE BENEFITS FOR MY CHILD AND OTHER PEOPLE IN THE FUTURE?

For people with undiagnosed moderate to severe intellectual disability, participation in this project may result in the establishment of a genetic diagnosis.

It is possible that this research may lead to the development of a face-matching program which has further research, not-for-profit or marketable potential. Any intellectual property will remain with Hunter New England Health and IMAGUS technology.

WHAT ARE THE POSSIBLE RISKS, SIDE EFFECTS, DISCOMFORT AND/OR INCONVENIENCES?

While we are developing the effectiveness of the face-matching program, it is possible that a match may not lead to a diagnosis or may raise an incorrect diagnosis. This could generate concern or disappointment for you as the parent/guardian.

It is possible that participation in the project may lead to the diagnosis of an inherited form of intellectual disability. This information may have implications for you, your children or other family members.

It is also possible that this information may be important in understanding the chance of having another child with the same condition. Referral to genetic counselling support can be arranged through your nominated doctor.

Even when parents have been searching for a diagnosis to explain the intellectual disability in their child, the actual time of diagnosis can be distressing. Support from your clinical geneticist and genetic counsellor will be available.

WHAT WILL BE DONE TO ENSURE THAT MY SON/DAUGHTER'S INFORMATION IS CONFIDENTIAL?

Any information collected for this research project will be treated as confidential. Information and photographs are stored securely away from the publically viewed website.

All uploaded photographs and data will be encrypted to maintain a high level of security. The information will be re-identifiable. This means that we will remove your name and your son/daughter's name and give the information a special code. Only the research team can match the names to their code number if necessary to do so.

The de-identified data generated by the computer vision software from the photograph is used for matching purposes. The original photograph and data (linked by code number) will be separately stored behind a firewall and password protected computer at Hunter Medical Research Institute. The decrypted images will be viewed by the FaceMatch research team.

When a high-probability match is made, de-identified images can be viewed within the secure FaceMatch site by:

1. Doctors nominated by the parents/guardians of the matched individuals.
2. Parents (only if parents of both matching individuals have consented to image sharing).

When we write or talk about the results of this project, information will be provided in such a way that your son/daughter cannot be identified.

WILL I BE INFORMED OF FACE-MATCHING RESULTS?

If your son/daughter has a high-probability face match with another individual within the database, information about this match will be sent to your nominated doctor who will contact you.

WHAT ARE THE DIFFERENT LEVELS OF CONSENT?

1. **CONSENT to participation in the FaceMatch Project as described in this information statement.**
2. **OPTIONAL CONSENT for sharing of de-identified images with other parents when a high-probability face-match is made.**

Images would only be shared within the secure FaceMatch site, and only if parents of both matching individuals have provided consent.

3. **OPTIONAL CONSENT to be contacted about future research projects.**

We would like you to consider allowing us to contact you and your doctor to discuss possible participation in future ethically approved projects.

4. **OPTIONAL CONSENT for development of a screening or diagnostic tool.**

It is possible that this research may lead to the development of a face-matching screening or diagnostic tool which has marketable potential. We would like you to consider allowing us to retain your child's de-identified image within the secure FaceMatch database for this purpose.

Thank you for taking the time to read this information statement.

If you would like more information about the project or if you need to speak to a member of the research team please contact the FaceMatch team at info@FaceMatch.org.au

If you have any concerns and/or complaints about the project, the way it is being conducted or your child's rights as a research participant, and would like to speak to someone independent of the project, please contact:

Nicole Gerrand
Manager, Research Ethics and Governance
Hunter New England Health Research Ethics Committee
HNELHD-HREC@hnehealth.nsw.gov.au
Please quote reference number 17/05/17/4.06

If you would like to participate in the project, please register yourself as a parent and when adding your child to the FaceMatch project, please indicate you have read and understood this information statement consent.